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Cystic Fibrosis (CF) is a common genetic disease. However, early detection can have a significant impact on improving the growth and nutrition of those affected with CF. The past year has been busy as the Hawai'i Newborn Metabolic Screening (NBMS) Program explored screening Hawai'i's newborns for CF.

The opportunity to add CF testing to the current NBMS panel presented itself when our contracted laboratory, the Oregon State Public Health Laboratory, decided to add CF to their panel of conditions. This decision was made in response to national recommendations published by the Centers for Disease Control and Prevention, the American College of Medical Genetics, the March of Dimes, the Cystic Fibrosis Foundation and the Health Resources and Services Administration.

The Hawai'i CF NBS Task Force met several times to discuss the benefits, limitations and policy issues involved in this endeavor. The Task Force report and recommendations in favor of adding CF to the NBS panel were presented at the annual NBS Advisory Committee meeting on September 14, 2006. The majority of the committee supported this recommendation, provided that the major program elements were in place before the anticipated start date of September 2007.

Newborn Screening for Cystic Fibrosis



While numerous administrative rules and program elements still need to be finalized, we are progressing in this multi-state and community effort to improve the health care of Hawai'i's newborns. Family, health provider, hospital, community and state representatives continue to be involved in the decision making process. We

will keep you posted about any new updates and information as they become available.

To read more about the importance of detecting babies with cystic fibrosis early, and the benefits of such a program in Hawai'i, you can download a copy of the CF NBS Task Force Report on our website.



Coordinator's Corner

We are pleased to announce that our local genetics community has expanded once again so that families in our state can have increased access to genetic services. We want to welcome Arthur Yu who is a new genetic counselor with the Fetal Diagnostic Center at Kapi'olani Medical Center for Women and Children. We also want to extend a warm welcome to Major Adam Kanis who is a clinical M.D. geneticist and as you can tell by his title, a military doctor. He is stationed at Tripler Army Medical Center. Please share your aloha with Arthur and Adam when you meet them.

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On October 17th, nearly 100 attendees attended our annual conference at the Japanese Cultural Center. This year's topic was, "Genetics For Your Practice: Putting The Pieces Together." The conference focused on developments in genetics at the local and national level.

Dr. Laurie Seaver opened the morning with her talk on "The Who, What, Why, and When of Genetics Referral" which Kirsty McWalter followed with "The Final W, Where." Both talks were embraced by participants as they learned about when and where to refer patients.

“Congratulations! Impressive collaboration and use of needs assessment.”

With referral tools in hand, participants heard updates on projects occurring in Hawai'i. Sylvia Au presented on the Western States Genetic Services Collaborative, a multi-state effort to increase access to genetic services. Lianne Hasegawa described the emerging use of telemedicine to increase access to services for neighbor island families. Participants were enthused to hear about the availability of the increased services and the use of new technology.

The morning closed with two educational talks on hearing loss and pharmacogenomics given by Elaine Marr and Sarah Scollon respectively. Participants felt these talks were very informative. Regarding the talk on hearing loss, one attendee found the information to be "useful for early intervention and our need to refer" while they felt the information on pharmacogenomics was "interesting and informative for a subject I knew little about."

The afternoon session opened with panel of speakers sharing their experience with genetic evaluations and counseling. Janet Brumblay walked us through what happens when a child has a positive newborn screening result. Allison Taylor and Michele Murray gave us insight into both prenatal and cancer genetic counseling. Finally, Amanda Kaahanui shared a parent's experience in navigating the health care system. Participants appreciated the real life examples and "enjoyed the parent perspective."



“This helps me to be able to give an idea to families of what to expect. Good overview of Hawai'i genetic resources.”

GENETICS FOR YOUR PRACTICE: PUTTING THE PIECES TOGETHER

The end of the afternoon encouraged participants to look at genetics through a different lens. Ken Takayama shared current updates on genetics legislation which was a new topic to many. One attendee commented, "This is so new to me. Thank you for including this presentation." Jennifer Bojanowski shared her personal experience in the New Orleans disaster recovery effort. Sharing the history of the use of genetics in missing person cases, she demonstrated a unique application of the science that listeners found "interesting and touching."

Finally, Sylvia Au closed off the day by asking the question, "Do you want to participate in genetic research?" She shared with us the desire of many researchers to begin large population studies to address the growing field of genomics. However, exploring this frontier raises many ethical, legal, and social questions. The discussion raised many interesting questions, engaged the crowd in thinking about the future of genetics and how they might participate in genetics research.

“The conference was great, well organized, and ran smoothly. Lots of info! I thought all sessions were helpful.”

The conference came together well and blended many different topics in the field. Participants learned how to practically apply genetics to their practice in Hawai'i as well as where genetics may be headed as we try to address the scope of genomics. We thank the many participants who came to join us to expand their knowledge of the field of genetics. We would also like to thank all of our guest speakers who took the time from their busy schedules to make the conference a success.

Ph.D

Research & Laboratory

Geneticists

This is the third installment of a continuing series of articles describing the various roles of genetics professionals. We hope the information will be useful for healthcare providers referring for genetic services, families who are referred for genetic services, and those considering a career in genetics.

In our last installment, we described the role of the clinical geneticists-M.D.s who focus on managing patients with genetic conditions. Now we will discuss Ph.D geneticists. Some may devote their time completely to basic research. Others may do more clinical laboratory research (e.g. pharmaceutical development, agriculture, etc.) or supervise the laboratories that offer clinical genetic testing. There are also M.D. geneticists in these roles, but this article will focus on the Ph.D geneticists.

Some Ph.D. geneticists work in basic research, building our knowledge about the genetics of natural biological processes. They have a solid background in the natural sciences, including math, chemistry, physics and biology. It usually takes 4 to 6 years to complete a Ph.D program and most graduates carry on their research as postdoctoral fellows for 2 to 4 more years. This type of geneticist may find a job as faculty at an academic institution or research institute.

The more clinically focused Ph.D geneticists work on creating new technologies and providing services in the areas of clinical medicine and legal or police work. They have similar training to the basic research geneticists. Their training, however, is more focused on molecular biology, cytogenetics, biochemical genetics, immunogenetics, and genetic technologies that can be applied for clinical or forensic use.

The work of Ph.D geneticists directly impacts families even though many may never have direct contact with the families. Examples include discovering the genes that cause cancer or other diseases and creating the technology to be able to test for those genes. Pharmaceutical developments revolve around creating drugs or therapies to treat genetic conditions or investigating how a person's genetic make-up may lead to different responses to drugs. Ph.D geneticists also use their knowledge to investigate how to breed new crop plants and healthier livestock. In legal or police work, forensic DNA analysis is done in a diverse number of settings including crime scene investigations, finding bacteria or other organisms that may pollute air, water, soil and food, identifying catastrophe victims, and in matching organ donors with recipients in transplant programs.

In Hawai'i, there are a number of research Ph.D geneticists mostly at the University of Hawai'i. They work in various areas, including evolution, biochemistry, biology, immunology, retrovirology, and agriculture. Presently, there is one practicing Ph.D geneticist certified by the American Board of Medical Genetics in Hawai'i. He is the director of the laboratory at the Queen's Comprehensive Genetics Center.

If you are interested in more information about these professions, please go to: American Society of Human Genetics www.ashg.org Genetics Society of America www.genetics-gsa.org

UPDATE



Teacher's Corner

We hope that teachers and students are settled in their classrooms after a busy summer! The Genetics Program also has been completing a pilot study and review of our "Teachers' Genetics Resource Kit" and receiving new DVDs from the Howard Hughes Medical Institute.

We would like to begin distributing resource kits to teachers on O'ahu. If you have a group of 5+ teachers at your school who teach genetics lessons (Biology, Life Sciences, Genetics, Biotechnology. etc.), then please contact us! We would be happy to schedule a short, 30-minute presentation at your school, including an overview of the materials in the kit, suggestions of how to use the kit, and kit distribution. If there are not enough teachers at your school, please consider gathering a group together from neighboring schools.

To receive the "Teachers' Genetics Resource Kit" and schedule our Genetics group at your school, please contact Kirsty McWalter (kirsty@hawaiiigenetics.org or 808-733-8387).



For more information, go to www.hawaiiigenetics.org

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ETIOLOGY OF HEARING LOSS PROJECT RESULTS

This September, the Hawai'i Genetics Program completed the "Etiology of Hearing Loss in Infants and Young Children" research project. This project, funded by the Centers for Disease Control and Prevention (CDC), sought to determine the causes of early-onset hearing loss in infants identified through the Newborn Hearing Screening Program (NBHSP). Families from Hawai'i, Utah and Rhode Island participated in the project.

During the research project, children received a full genetics evaluation by Dr. Melanie Manning, a geneticist from Stanford University. They also received testing for four genes that can cause hearing loss in newborns and children: Connexin 26, Connexin 30, and two genes in the mitochondria. This type of hearing loss is non-syndromic, meaning that the hearing loss is not associated with other medical issues.

A total of 17 children from O'ahu, Maui, and the Big Island were evaluated. Of the twelve children who have received genetic test results, five were found to be positive for Connexin 26 mutations. This means that their hearing loss is caused by changes in their Connexin 26 gene. Five others are still awaiting their test results.

Data gathered from this research project will be combined with similar data collected from Utah and Rhode Island. Analysis of the results will provide us with a better understanding of the causes of hearing loss in infants identified by the NBHSP. This will help improve follow-up genetic services for these children. We would like to thank all of the families who participated in this project! It is because of their participation that we are able to discover new information and provide better services to future generations. Mahalo!

A
Parent's Guide
to the
Etiology of
Hearing Loss
Research
Project

